



BJMG is an open access, peer-reviewed journal. All articles in the journal are immediately and permanently available online for all to read and use free of charge.

BJMG is published twice a year.

Indexed or abstracted in: EMBASE (the Excerpta Medica database), Elsevier Biobase (Current Awareness in Biological Sciences), Elsevier GeoAbstracts, PubMed Central, Scopus, Chemical Abstracts (CA), Science Citation Index Expanded, SCImago (SJR) Journal Citation Reports/Science Edition, De Gruyter, Sciendo, Academic Open Access Publisher.

BJMG is covered by Thomson Reuters services.

EDITORS

Plaseska-Karanfilska D (Skopje), Toncheva DI (Sofia)

PAST EDITOR

Efremov GD (Skopje)

EDITORIAL BOARD

Apak M (Istanbul)	Geraedts J (Maastricht)	Nakamara Y (Tokyo)
Avent N (Plymouth)	Goossens M (Paris)	Pavelic K (Zagreb)
Baranov V (St Petersburg)	Kanavakis E (Athens)	Peterlin B (Ljubljana)
Barisic I (Zagreb)	Keohavong P (Pittsburg)	Stevanovic M (Belgrade)
Basak N (Istanbul)	Kocova M (Skopje)	Todorova A (Sofia)
Czeizel A (Budapest)	Kremenski I (Sofia)	Tomlinson I (London)
Cooper D (Cardiff)	Kucinskas V (Vilnius)	Vella F (Saskatoon)
Dimovski AJ (Skopje)	Khusnutdinova E (Ufa)	Wajcman H (Paris)
Dörk T (Hannover)	Labie D (Paris)	Yapiyakis C (Athens)
Efremov DG (Trieste)	Liehr T (Jena)	Zoraqi G (Tirana)
Fryns JP (Leuven)	Lungeanu A (Bucharest)	

EDITORIAL ASSISTANTS

Sukarova Stefanovska E, Skopje
Carver M.F.H, Evans, GA, USA
Janeska K, Skopje

COVER DESIGN

Bitrakova-Grozdanova V, Skopje

EDITORIAL OFFICE

Macedonian Academy of Sciences and Arts, Bul. Krste Misirkov 2, POB 428
1000 Skopje, Republic of North Macedonia.
Phone: + 389 2 3235 411, fax: + 389 2 3115 434, E-mail: icgib@manu.edu.mk
Web page: www.bjmg.edu.mk

PUBLISHER



BJMG is published by Macedonian Academy of Sciences and Arts

Printed by: Art Print Studio DOOEL, Skopje, Republic of North Macedonia

Front cover page: Sarcophagus with Hippolytus and Phaedra, marble, Salona, Manastirine, early 4th century, AM, Split, Croatia.

Back cover page: Mosaic fragment with a motif of Traditio legis, St. Maria Formosa, Pola, mid 6th century, AMI Pula, Croatia.

CONTENTS

Review Articles

GENETIC SPECTRUM OF NEONATAL DIABETES Kocova M.....	5
---	---

Original Articles

THE UTILITY OF WHOLE EXOME SEQUENCING IN DIAGNOSING PEDIATRIC NEUROLOGICAL DISORDERS Muthaffar OY.....	17
ARRAY-COMPARATIVE GENOMIC HYBRIDIZATION RESULTS IN CLINICALLY AFFECTED CASES WITH APPARENTLY BALANCED CHROMOSOMAL REARRANGEMENTS Satkin NB, Karaman B, Ergin S, Kayserili H, Kalelioglu IH, Has R, Yuksel A, Basaran S	25
DO WE USE METHYLATION OF <i>NFATC1</i> AND <i>FOS</i> GENES AS A BIOMARKER FOR POSTMENOPAUSAL OSTEOPOROSIS? Kalkan R, Tosun O.....	35
INFLUENCE OF POTENTIAL GENE POLYMORPHISMS ON PROPOFOL DOSAGE REGIMEN IN PATIENTS UNDERGOING ABDOMINAL HYSTERECTOMY Ivanov E, Sterjev Z, Budic I, Nojkov J, Karadzova D, Sivevski A	41
INVESTIGATION OF THE RELATIONSHIP OF <i>TNFRSF11A</i> GENE POLYMORPHISMS WITH BREAST CANCER DEVELOPMENT AND METASTASIS RISK IN PATIENTS WITH <i>BRCA1</i> OR <i>BRCA2</i> PATHOGENIC VARIANTS LIVING IN THE TRAKYA REGION OF TURKEY Özdemir K, Gürkan H, Demir S, Atli E, Özen Y, Sezer A, Tunçbilek N, Çicin İ.....	49
PROS AND CONS FOR FLUORESCENT <i>IN SITU</i> HYBRIDIZATION, KARYOTYPING AND NEXT GENERATION SEQUENCING FOR DIAGNOSIS AND FOLLOW-UP OF MULTIPLE MYELOMA Ikbal Atli E, Gurkan H, Onur Kirkizlar H, Atli E, Demir S, Yalcintepe S, Kalkan R, Demir AM.....	59
EVALUATION OF METHYLATION PROFILES OF AN EPIDERMAL GROWTH FACTOR RECEPTOR GENE IN A HEAD AND NECK SQUAMOUS CELL CARCINOMA PATIENT GROUP Mutlu M, Mutlu P, Azarkan S, Bayır Ö, Öcal B, Saylam G, Korkmaz MH.....	65
EVALUATION OF MicroRNA-124 EXPRESSION IN RENAL CELL CARCINOMA Çaykara B, Öztürk G, Alsaadoni H, Ötünçtemur A, Pençe S	73

Case Reports

**FETAL CYSTIC HYGROMA ASSOCIATED WITH TERMINAL 2p25.1 DUPLICATION AND
TERMINAL 3p25.3 DELETION: CYTOGENETIC, FLUORESCENT *IN SITU* HYBRIDIZATION
AND MICROARRAY FAMILIAL CHARACTERIZATION OF TWO DIFFERENT CHROMOSOMAL
STRUCTURAL REARRANGEMENTS**

Stipoljev F, Barbalic M, Logara M, Vicic A, Vulic M, Zekic Tomas S, Gjergja Juraski R79

**FAMILIAL NON-AUTOIMMUNE HYPERTHYROIDISM IN FAMILY MEMBERS ACROSS
FOUR GENERATIONS DUE TO A NOVEL DISEASE-CAUSING VARIANT
IN THE THYROTROPIN RECEPTOR GENE**

Malej A, Avbelj Stefanija M, Bratanič N, Trebušak Podkrajšek K.....87

**BILATERAL RENAL ANGIOMYOLIPOMAS AND SUBEPENDYMAL GIANT CELL
ASTROCYTOMA ASSOCIATED WITH TUBEROUS SCLEROSIS COMPLEX:
A CASE REPORT AND REVIEW OF THE LITERATURE**

Rambabova Bushljetik I, Lazareska M, Barbov I, Stankov O, Filipce V, Spasovski G93

**TWO NOVEL *CEBPA* MUTATIONS IN A TURKISH PATIENT
WITH ACUTE MYELOID LEUKEMIA**

Tokgun PE, Alay MT, Atli Tekin S, Güler N, Tokgun O, Demiray A, Karagenc N, Durak T, Celik B, Akca H.....99

**CASE REPORT OF A SUCCESSFUL PREGNANCY IN A CYSTIC FIBROSIS PATIENT
WITH THE c.1521_1523delCTT/c.3718-2477C>T GENOTYPES**

Spasova VL, Koleva LI, Toncheva DI, Karamisheva VI.....103

**OSTEOPETROTRICKETS IN AN INFANT WITH COEXISTENT
CONGENITAL CYTOMEGALOVIRUS INFECTION**

Katsafiloudi M, Gombakis N, Hatzipantelis E, Tragiannidis A.....107